

Genetic Testing Registry (GTR)
Draft Document Release
February 2011

The draft documents described below represent working versions of design and development elements for GTR. As such, they should be considered subject to change without notice, and they may not fully or accurately reflect the final version of GTR when it goes into production.

The documents incorporate feedback from a number of GTR stakeholder meetings, two advisory groups, and visits and communications from interested parties. NCBI is building a prototype database and sample XML that reflect these documents, as well as a prototype website to display records containing this information; these prototypes will allow GTR to begin testing real data and real life scenarios.

The GTR design is not final, and it is expected that there will be further changes and refinements based on suggestions, test cases, and the insights that come from building the first version. However, the design is sufficiently developed that it is ready for specific comments.

Constructive feedback is encouraged and welcomed. Please email gtr@ncbi.nlm.nih.gov to make comments or arrange a meeting.

This release of documents contains the following files:

README - this document

GTR Design Considerations - A brief overview of the main components and design considerations for the GTR, intended to aid in seeing the forest instead of the trees.

Proposed GTR Field Definitions - A comprehensive listing of the data fields in GTR; these are the trees. This document is not a formal schema but instead meant to be an explanation, field by field, of the proposed data elements in GTR. Not every laboratory or test would use all the fields.

Example files - There are 5 examples, each in a separate file, which show how the fields might be used for specific tests. Any one example does not use all the fields in the Field Definitions document, but collectively the examples show ALL the fields that might be populated for specific types of tests. Please note that these are simply the fields for the underlying database and do NOT reflect how these tests would be presented on the actual GTR web site. On the working site, many of these fields would be summarized into condensed tables or would only appear on supplementary web pages, to make the site conducive to quick exploration of multiple tests.

XML example - An XML example is provided, although the details of the XML are still in early development. This document is intended to give technically minded people an idea of how the XML may represent the Field Definitions. There are still many issues to be resolved on the specifics of bulk uploading or downloading GTR data, and NCBI invites conversation on these aspects of GTR. Although the XML issues are not completely separate from the Field Definitions issues, we believe it will be more effective to have this discussion separately. Again, please contact gtr@ncbi.nlm.nih.gov to participate in this side of the discussion. More XML examples will be made available to this group.